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A recessively inherited windmill-vane camptodactyly/ichthyosis syndrome

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SUMMARY A brother and sister are described with a syndrome, not previously reported, of distal arthrogryposis, facial immobility, and generalised ichthyosis.

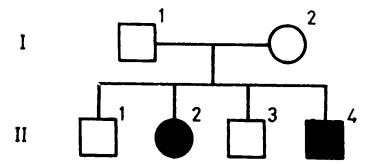

FIG 1 *Family pedigree.*

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In their recent review of distal arthrogryposis, Hall *et al*[1] proposed two major categories for their patients; type 1 with isolated contractures and type 2 where additional abnormalities are present. The best known specific diagnosis in the latter group is the 'whistling face' or Freeman-Sheldon syndrome, characterised by camptodactyly with ulnar deviation, vertical talus, and facial immobility. The condition

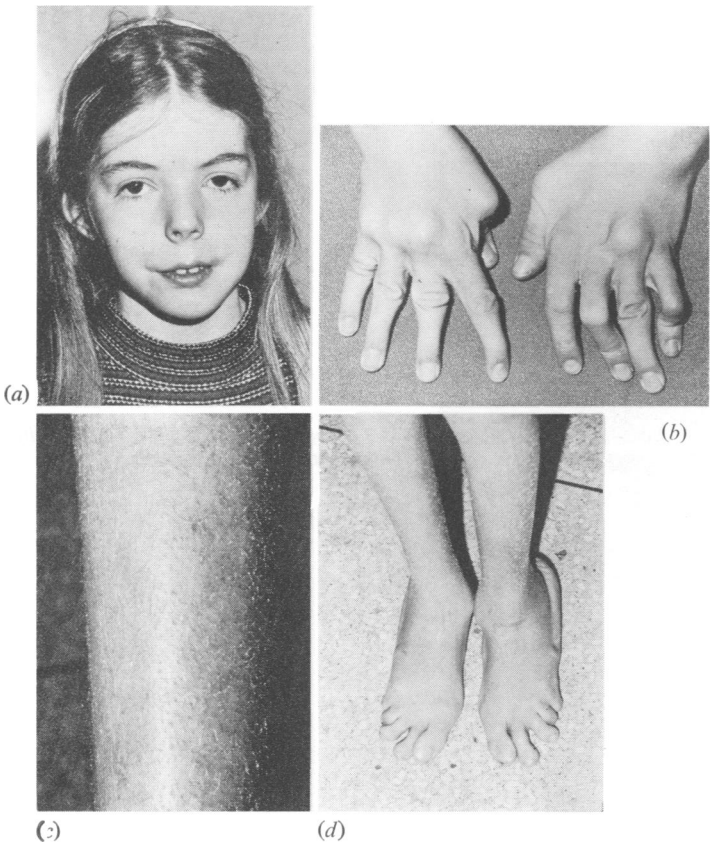

FIG 2 *Appearance of II.2. (a) Immobile face with limitation of mouth opening. (c) Generalised ichthyosis. (b, d) Camptodactyly and lateral deviation of digits.*

behaves as an autosomal dominant, although there has been a single report of Freeman-Sheldon syndrome in sibs[2] raising the possibility of heterogeneity. The present report describes a sib pair with many features of Freeman-Sheldon syndrome but with, in addition, ichthyosis. These children may be suffering from a distinct autosomal recessive syndrome.

Case reports

The affected sibs were among four children of unrelated English parents (fig 1).

CASE 1

II.2 (470576) was born by extended breech delivery at term on 13.4.73. Fetal movements were normal but liquor volume was markedly reduced. Fig 2 illustrates the important clinical features.

Her principal skeletal disorders are: limitation of hip movements, vertical talus, calcaneovalgus deformity, ulnar deviation of the fingers, camptodactyly affecting all limbs, a thoracic scoliosis developing in late childhood, and spina bifida on *x*-ray at S1. The severity of the camptodactyly has increased with time (fig 2b,d).

Neurological examination in early childhood revealed marked motor delay. She was slow to vocalise, as were the normal sibs, but now has no significant intellectual retardation. General muscular hypoplasia became obvious in mid-childhood but has not shown signs of progression.

The skin showed quite severe generalised ichthyosis involving flexures (fig 2c). The principal facial features were a broad prominent forehead, wide set eyes, taut facial skin with limitation of mouth opening, and lack of facial expression (fig 2a).

CASE 2

The second affected sib, II.4 (522460), exhibited the same features but to a lesser degree (fig 3). Pregnancy was associated with reduced fetal movement and liquor volume was very small. Birth weight was 3·4 kg and delivery was with cephalic presentation and extended legs. Again arthrogryposis was evident. The camptodactyly, ulnar deviation of hands, valgus foot deformity with vertical talus (fig 4), and reduced facial mobility became evident though were less

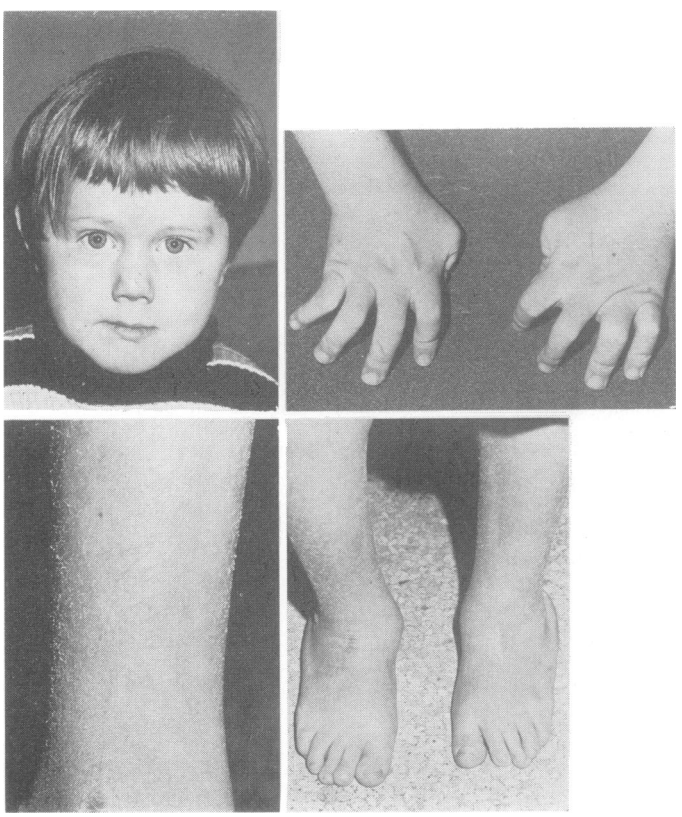

FIG 3 *Appearance of younger sib II.4 with similar but less severe features.*

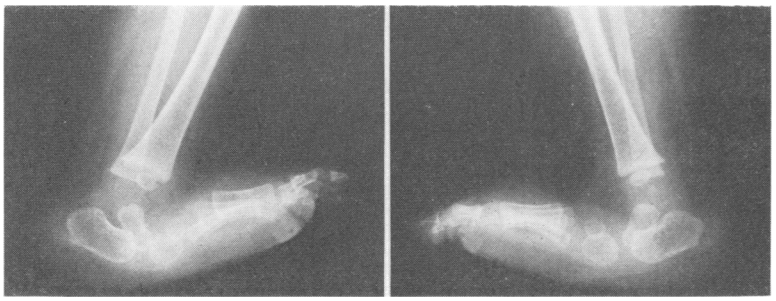

FIG 4 *Lateral radiograph of feet of II.4. Note vertical talus.*

severe. Muscular hypotonia was noted but was not progressive. Creatine kinase estimation was normal. Development gave cause for concern but, apart from delayed motor milestones, intellectual development was comparable to the healthy sibs. As with II.2, the ichthyosis was evident in infancy becoming worse with time and involving flexures. Both sibs required emollient skin applications from early childhood.

Discussion

The elder sib in this report was initially considered to have Freeman-Sheldon syndrome. However, the birth of an affected brother in the presence of phenotypically normal parents necessitated that the diagnosis be reviewed.

Three recessive conditions are known to include distal arthrogryposis: Tel-Hashomer,[3] Schwartz-Jampel,[4] and Guadalajara[5] syndromes. The Schwartz-Jampel syndrome can be excluded since ulnar deviation is not a feature and the blepharophimosis, characteristic of that syndrome, was not present in these sibs. Camptodactyly with intrauterine growth retardation, known as the Guadalajara camptodactyly syndrome, has as its main features agenesis of the frontal-ethmoidal sinuses, an abnormal position of the left canine, prognathism, depressed lower half of the sternum, fibular hypoplasia, mild mental retardation, and metaphyseal broadening. These associated features were not found in the sibs presented. Goodman's Tel-Hashomer syndrome has many features in common with the sibs in this report: short stature, brachycephaly with prominent forehead, hypertelorism, small mouth, increased philtral length, and poor muscle development. Ichthyosis has not been reported. All four of the patients of Goodman *et al*[3] had malformed toes, two had dislocated radii, and two had club feet. In the two previous reports one set of parents was Jewish Moroccan and the other was Arab Bedouin.

Alves and Azevedo[2] reported two sibs with flattening of the supraorbital ridges, moderate blepharophimosis, ptosis, antimongoloid eye slant, a parrot-like nose, and irregular dimples of the skin over the chin, strongly suggestive of the 'whistling face' syndrome. However, there was no camptodactyly or ulnar deviation in either the hands or the feet. The sibs in the present report can be distinguished from those described by Alves and Azevedo, and from those reported in association with the other syndromes discussed, by the presence of congenital ichthyosis.

When counselling the normal parents of a child with features of Freeman-Sheldon syndrome, the possibility of a rare recessive form should be considered. If, in addition to the facial and skeletal anomalies, the child manifests congenital ichthyosis, it would seem prudent to regard this as a provisional autosomal recessive condition.

References

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